



NEFL gene

neurofilament light

Normal Function

The *NEFL* gene provides instructions for making the smallest protein component (the light subunit) of neurofilaments, which are essential for normal nerve cell function. Neurofilaments are assembled from light, medium, and heavy subunits. They form a structural framework that helps to define the shape and size of nerve cells. Cross-linking or bridging between neurofilaments maintains the diameter of the fiber, or axon, that extends from a nerve cell. Maintaining the proper axon diameter is essential for the transmission of nerve impulses.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

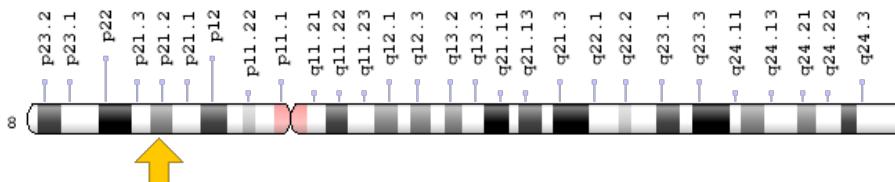
Researchers have identified more than 10 *NEFL* gene mutations that cause two forms of Charcot-Marie-Tooth disease known as type 1F and type 2E. Most *NEFL* gene mutations change single protein building blocks (amino acids) in the neurofilament light subunit. Other mutations delete or duplicate part of the *NEFL* gene, which alters the instructions for making the neurofilament light subunit. *NEFL* gene mutations probably alter the size or shape of neurofilament light subunits.

Altered neurofilament light subunits likely disrupt the assembly of neurofilaments or their transport to the axon. These disruptions may cause abnormalities in axons and impair the transmission of nerve impulses. Abnormalities in axons are a sign of type 2E Charcot-Marie-Tooth disease. In some cases, a reduced axon diameter may result in decreased nerve signal transmission speed, which is characteristic of type 1F Charcot-Marie-Tooth disease.

Chromosomal Location

Cytogenetic Location: 8p21.2, which is the short (p) arm of chromosome 8 at position 21.2

Molecular Location: base pairs 24,950,955 to 24,956,869 on chromosome 8 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT1F
- CMT2E
- neurofilament triplet L protein
- neurofilament, light polypeptide
- neurofilament, light polypeptide 68kDa
- NF68
- NFL
- NFL_HUMAN
- PPP1R110

Additional Information & Resources

Educational Resources

- Molecular Cell Biology (fourth edition, 2000): Intermediate Filaments
<https://www.ncbi.nlm.nih.gov/books/NBK21560/>
- The Cell A Molecular Approach (second edition, 2000): Intermediate Filament Proteins
<https://www.ncbi.nlm.nih.gov/books/NBK9834/>

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 1
<https://www.ncbi.nlm.nih.gov/books/NBK1205>
- Charcot-Marie-Tooth Neuropathy Type 2
<https://www.ncbi.nlm.nih.gov/books/NBK1285>
- Charcot-Marie-Tooth Neuropathy Type 2E/1F
<https://www.ncbi.nlm.nih.gov/books/NBK1187>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NEFL%5BTIAB%5D%29+OR+%28%28NFL%5BTIAB%5D%29+OR+%28neurofilament+triplet+L+protein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- NEUROFILAMENT PROTEIN, LIGHT POLYPEPTIDE
<http://omim.org/entry/162280>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NEFL.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NEFL%5Bgene%5D>
- HGNC Gene Family: Intermediate filaments Type IV
<http://www.genenames.org/cgi-bin/genefamilies/set/611>
- HGNC Gene Family: Protein phosphatase 1 regulatory subunits
<http://www.genenames.org/cgi-bin/genefamilies/set/694>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7739
- Inherited Peripheral Neuropathies Mutation Database
<http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=8>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4747>
- UniProt
<http://www.uniprot.org/uniprot/P07196>

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